Prevalence of BRCA1 185delAG, BRCA1 5382insC and BRCA2 617delT Mutations and their Association with Breast Cancer in Jordanian Females

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ABSTRACT

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Breast cancer (BC) is the most common cancer occurring among women; and represents about 36.8% of all cancers in Jordanian women. Many environmental and genetic factors contribute to BC, but BRCA1 and BRCA2 hereditary BC was not fully studied in Jordan.

In this study, the prevalence of the founder mutations BRCA1 185delAG, BRCA1 5382insC and BRCA2 6174delT have been studied in 300 samples representing the Jordanian population. In addition, the associations of each of these mutations with BC were tested in a case – control study that included 200 females affected with breast cancer and 200 age-matched female controls collected from the Northern and middle parts of Jordan. Written informed consent was obtained from each of the
participating individuals. DNA was isolated from peripheral blood using OMIGA kit. The detection of the three mutations BRCA1 185delAG, BRCA1 5382insC and BRCA2 6174delT was accomplished using a mutagenically separated PCR. BRCA1 185delAG and BRCA2 6174delT were not detected in this study, while BRCA1 5382insC was detected in the cases in one heterozygote with allele frequency of 0.25% in the cases and 0.1% in the whole Jordanian population. This is the first study concerning the prevalence of BRCA1 and BRCA2 mutations in Jordan. The low frequencies of BRCA1 and BRCA2 genes in the Jordanian population are similar to other studies that showed low frequency of these three mutations in other populations. In conclusion, our results indicated that most of BC cases in Jordan may be due to alterations in other genes and/ or due to environmental conditions.